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PERIANAL LEIOMYOMA INVOLVING THE ANAL SPHINCTER.

Editor,

Leiomyomas are benign soft tissue tumours of mesenchymal origin and can develop wherever smooth muscle is present. Their pathogenesis remains obscure. Deep soft tissue leiomyomas are rare and are further classified as somatic and retroperitoneal. Whereas the former have a predilection to occur in extremities (usually in the thigh) the latter usually occur in the pelvic retroperitoneum[1]. We report a case of perianal leiomyoma stretching the muscle fibres of the external sphincter. Reports of perianal leiomyomas are sparse in the literature. Features of deep soft tissue leiomyomas, anal leiomyomas and their management are discussed.

Clinical background: A 45-year-old female presented with a history of a painless swelling in the perianal region for 18 months, gradually increasing in size. Clinical examination revealed a 30mm diameter extrasphincteric swelling in the rectovaginal septum. Endoanal ultrasonography showed a soft tissue mass related to the anterior and lateral wall of the anal canal over its entire length. Although the mass appeared to be entirely outside the external sphincter complex there was a suspicion of sphincter involvement anteriorly. The lesion was well defined and homogeneous in texture with an intermediate to low signal intensity on T2 weighed magnetic resonance imaging (Figure 1). Fat saturation (FAT SAT) & Short Tau Inversion Recovery (STIR) sequences suggested that the lesion displaced rather than infiltrated the sphincter. There was loss of visualisation of the lower subcutaneous and superficial components of the external sphincter with a suspicion of extension to the deeper component of the anal sphincter.

An elective excision was performed with a circumanal incision. Sphincter fibres were stretched over the surface of the lesion. Complete extra capsular dissection of the lesion was performed in continuity. Sphincter fibres were divided and repaired with 2'0 PDS.

Macroscopically, the tumour was solid and well circumscribed with a whorled white cut surface without gross cystic

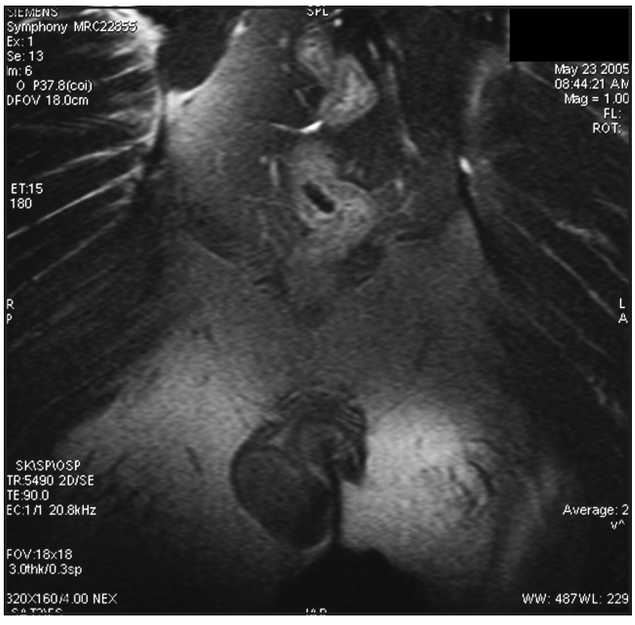

Fig 1. MR sequence with T2 weighting with fat saturation demonstrating an ovoid shaped low signal mass in relation to the right side of anal canal displacing the fibres of external sphincter.

degeneration or necrosis. The tumour measured 65mm in diameter. Histological examination revealed a circumscribed smooth muscle tumour consisting of interlacing fascicles of bland spindle cells admixed with focal areas of myxohyaline stroma. There was no cytological atypia, abnormal mitotic activity or necrosis. Only one or two mitoses were identified in the sections examined. Immunohistochemistry demonstrated strong positivity for smooth muscle markers desmin (Figure 2) and actin. Positivity for estrogen and progesterone receptors was also noted. CD117 was negative. Two months after the surgery, the patient has no incontinence with good sphincter tone.

Discussion: First described by Virchow in 1854, leiomyomas are benign soft tissue tumours that arise from smooth muscle accounting for 3.8% of all benign soft tissue tumours[1]. Kloepfer originally noted a hereditary syndrome characterised by multiple leiomyomas in 1958. Leiomyomata can develop

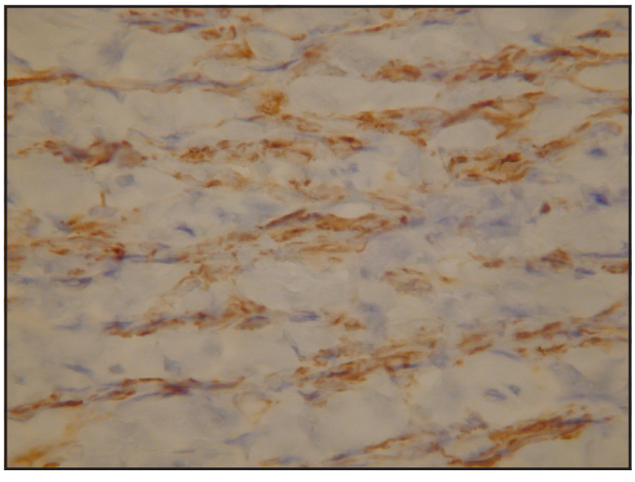

Fig 2. Immunohistochemical staining for Desmin (immunoperoxidase, x 250)

wherever smooth muscle is present, the commonest site being the uterine myometrium.

Leiomyomas are classified into superficial pilar, genital, angioleiomyoma and deep varieties. The pathological features of deep soft tissue leiomyomas were first described by Kilpatrick et al[2] and Billings et al[3]. They are further categorised as somatic and retroperitoneal types[2, 3]. Whereas somatic soft tissue leiomyomas affect the sexes equally with predilection to the extremities (usually in the thigh), retroperitoneal leiomyomas occur preferentially in women during the peri-menopausal period, usually in the pelvic retroperitoneum[1].

Somatic leiomyomas most often present as a localised mass. In addition, perianal leiomyomas tend to cause discomfort in the seated posture and also during defecation. Gastrointestinal symptoms such as constipation and bleeding are uncommon. The perianal region is a rare site and proximity to the sphincter complex can have considerable implications for operative management[1]. Examination reveals small rubbery to large lobulated firm lesions with intact mobile mucosa in those with endoluminal extension.

Macroscopically, deep soft tissue leiomyomas tend to be well defined and are usually surrounded by a fibrous pseudocapsule. They tend to be larger than superficial leiomyomas since they tend to remain occult by virtue of their site. Histologically, somatic soft tissue leiomyomas are composed of interlacing bundles of mature smooth muscle cells with abundant eosinophilic cytoplasm which, by definition, lack atypia and necrosis and are mitotically inactive (<1 mitoses/50 high power fields). Myxohyaline degeneration and regressive changes are constant features. Foci of dystrophic calcification are commonly present.

Unlike somatic soft tissue leiomyomas, 20% of retroperitoneal or abdominal leiomyomas display low levels of mitotic activity (<5 mitoses / 50 HPF[4] or <1-10 /50 HPF[3]). Leiomyomas of the anal canal arise in the muscle coat or less commonly in the muscularis mucosae. They grow slowly and the anoderm usually remains intact. Within the rectum and anal canal, leiomyomas can adopt different growth patterns, namely endoluminal, intramural or extraluminal. Most leiomyomas of the large bowel and rectum grow endoluminally whereas tumours of the anal canal tend to grow away from the lumen[5]. Sometimes they grow in both directions, forming an 'hour glass'.

Many tumours previously regarded as leiomyomas of the gastrointestinal tract are now considered as GISTs. Although the incidence of anal canal GIST is low (<2%), 10 to 30% of GISTs are malignant[6]. GISTs are more common than other mesenchymal tumours of the gastrointestinal tract except in the oesophagus where leiomyomas predominate. GISTs are differentiated from leiomyomas on the basis of immunohistochemical staining patterns including positivity for CD117, CD34, and smooth muscle actin and are usually negative for desmin that tends be expressed by the latter[6]. Currently the best indicator of malignancy in GISTs is the presence of invasion of adjacent organs or metastatic disease seen on imaging or at surgery.

The treatment of choice for anal canal leiomyomas and low grade GISTs is excision. Sphincter preservation should be possible. High grade GISTs require wide excision that might lead to considerable sphincter damage[5]. Unlike GISTs, deep soft tissue leiomyomas have a low recurrence rate if local excision is complete[2, 4] Deep soft tissue lesions that lack atypia, necrosis and mitotic activity and retroperitoneal lesions with <10 mitoses / 50 HPF can be labelled benign with reasonable confidence expecting a good outcome. Lesions falling outside this criteria and not obviously malignant (characterised by atypia and mitoses) should be considered as tumours of uncertain malignant potential in which case a regular follow up is advised[5, 6].

Conclusion: Deep soft tissue leiomyomas in the perianal region are rare. They may mimic anal leiomyomas and GISTs when they extend close to the sphincter. Despite the similarity in clinical presentation, histological features and prognosis, it is important to identify GISTs based on immunohistochemistry for CD117 since those with malignant potential require regular follow up and many of these tumours will benefit from imatinib mesylate, an inhibitor of the c-kit tyrosine kinase receptor. As with all spindle cell neoplasms, meticulous histopathological attention to the presence of significant mitotic activity, atypia and necrosis is essential since these factors would suggest potential malignant behaviour in which case a more radical surgical excision and follow up would be warranted.

The authors have no conflict of interest

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